Pediatr Radiol (2002) 32: 373–375
DOI 10.1007/s00247-002-0662-2

CASE REPORT

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Prenatal diagnosis of hypochondrogenesis using fetal MRI: a case report

Received: 2 April 2001
Accepted: 30 November 2001
Published online: 9 March 2002

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Abstract We describe the successful prenatal diagnosis of hypochondrogenesis by MRI. Fetal MR findings were the presence of a conspicuous cartilaginous structure in the basioccipital region, ill-defined ossification of the cervical vertebral bodies, hypoplastic thorax, retarded ossification of the pubic bones, and broad, short long bones. In contrast, fetal US revealed only the presence of short long bones. MRI accurately delineated the axial skeleton in this case and is an effective clinical tool for diagnosing skeletal dysplasias in utero.

Keywords MRI · Fetus · Hypochondrogenesis

Introduction

An accurate prenatal diagnosis of a malformed fetus not only facilitates prenatal decision making, but also assists in the provision of appropriate postnatal patient care. We recently examined by MRI a fetus with clinical features of hypochondrogenesis – severe spondyloepiphyseal dysplasia congenita (SEDc) spectrum. The clinical and imaging features of the patient are described and the potential role of fetal MRI in the antenatal management of skeletal dysplasias is discussed.

Case report

The pregnant mother was referred at 30 weeks' gestation because fetal US had revealed polyhydramnios and abnormally short limbs. Re-examination of the fetus by US confirmed the presence of short limbs (-7.8 SD), but did not reveal any other abnormal findings. Fetal MRI using the HASTE sequence at 32 weeks' gestation revealed the presence of a conspicuous cartilaginous structure in the basioccipital region, ill-defined ossification of the cervical vertebral bodies, hypoplastic thorax, retarded ossification of the pubic bones, and broad, short long bones (Figs. 1, 2). Given these findings, a diagnosis of hypochondrogenesis was made.

The mother delivered the female baby at 40 weeks' gestation by caesarean section. Birth weight was 2,944 g. Postnatal radiographs confirmed the diagnosis of hypochondrogenesis (Fig. 3). Shortly after birth, the infant was intubated because of severe respiratory distress as a result of hypoplastic lungs. She survived respiratory failure and mechanical ventilation was continued until the age of 7 months, at which time she died suddenly of unexplained severe hyperthermia and tachycardia. Autopsy disclosed absent enchondral ossification of cervical vertebral bodies and pubic bones.

Discussion

Achondrogenesis type II, hypochondrogenesis and SEDc constitute a continuous clinical spectrum of dis-

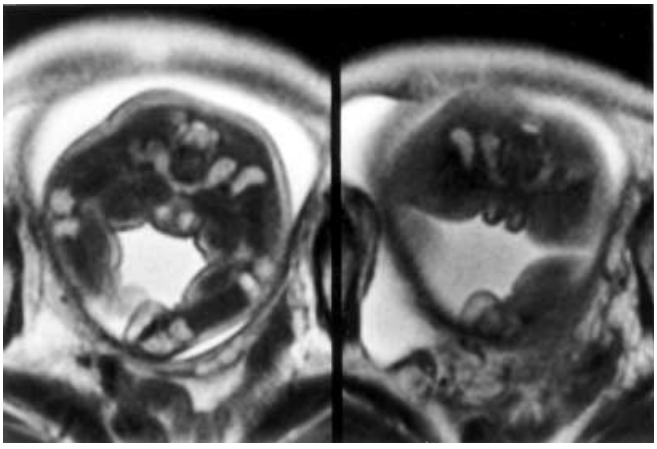

Fig. 1. Fetal MRI. Ossification of pubic bones is absent. The long bones of the lower extremities are markedly shorter than normal

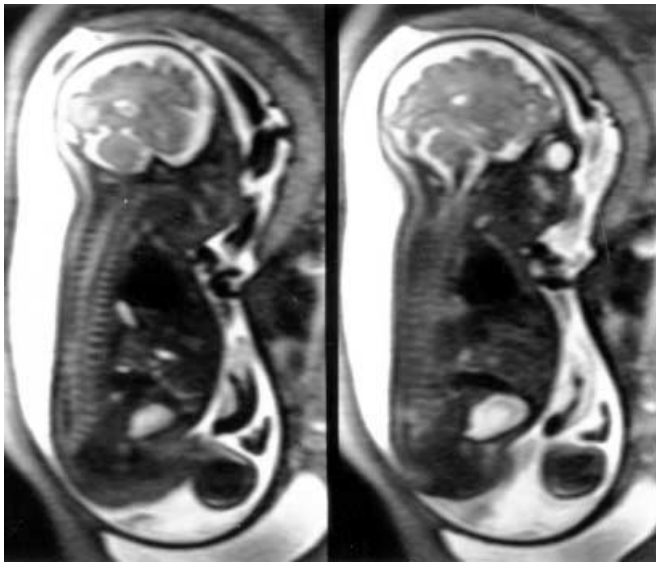

Fig. 2. Fetal MRI. Ossification of cervical vertebral bodies is absent. The ossification defect in the occiput is easily identifiable

orders, which are caused by mutations of the type II collagen gene [1]. At the severest end of the spectrum, achondrogenesis type II is characterised by the absent ossification of vertebral bodies and pubic and ischial bones, as well as concave inferior and medial margins of the ilia and metaphyseal ends. The mildest end, typical SEDc, shows ovoid or pear-shaped vertebral bodies, absent pubic ossification and absent epiphyseal ossification.

An intermediate phenotype is hypochondrogenesis – severe SEDc. The radiological characteristics of the hypochondrogenesis spectrum include an ossification defect behind the foramen magnum and the absence of cervical vertebral body ossification, as well as skeletal changes identical to those of typical SEDc. Shortening and metaphyseal modifications of the long bones are generally more conspicuous in hypochondrogenesis spectrum than in typical SEDc [2]. In the present patient, fetal MRI defined most of these radiological characteristics, and virtually identified the diagnosis, although a

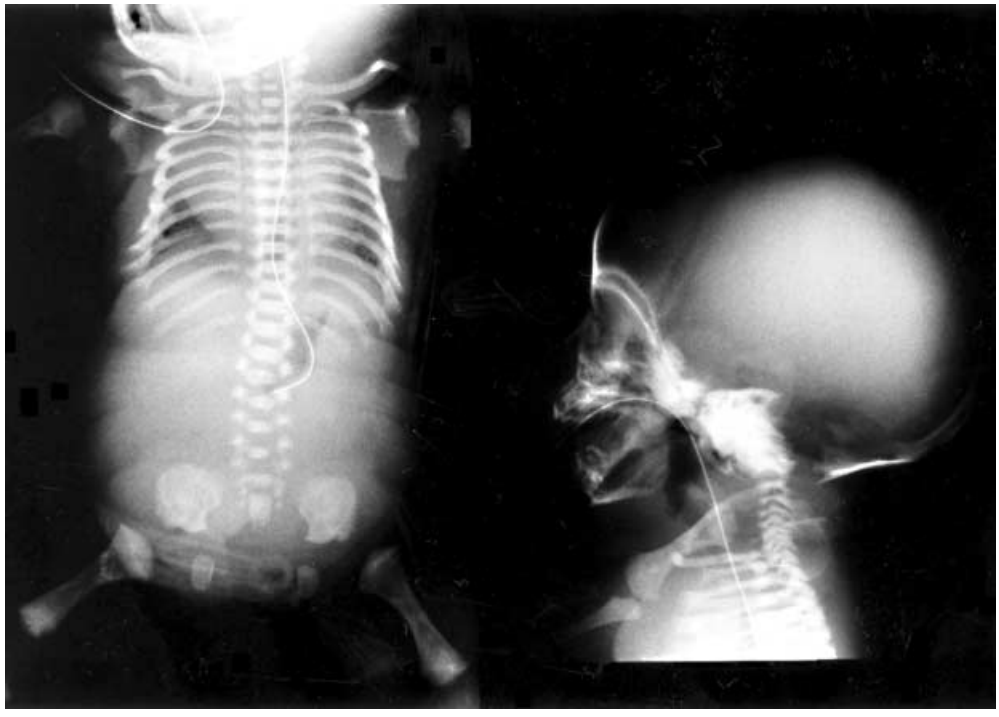

Fig. 3. Postnatal radiographs showing the characteristic radiological features of hypochondrogenesis. The long bones of the lower extremities are remarkably short compared with the upper extremities. Ossification of pubic bones and cervical vertebral bodies is absent. The ossification defect of the occiput is clearly visualised

mild form of platyspondylic lethal chondrodysplasia could not be completely excluded.

Fetal US currently plays the most important role in the prenatal diagnosis of skeletal dysplasia [3]. However, despite rapid technological advancements in US, its capability to visualise the fetal skeleton in sufficient detail is limited [4]. Prenatal US diagnosis of achondrogenesis type II has been successful because of the extreme shortening of the limbs and ossification defects of the spine. Nevertheless, the hypochondrogenesis spectrum has not previously been precisely diagnosed prenatally. Retarded ossification of the ischial and pubic bones, which points to hypochondrogenesis, cannot be detected by US, but can be demonstrated by MRI.

High-quality fetal radiography can facilitate a prenatal diagnosis of skeletal dysplasias. However, its routine use is impractical in prenatal diagnosis, particularly because of difficulties in positional control of fetuses, let alone potential hazards of irradiation. The ability of MRI to produce sectional images is advantageous and its capability to delineate cartilage as well as bone may provide additional information, such as the degree of ossification defects, that is not obtainable by radiography.

The relatively long imaging acquisition times of MRI often produce image degradation from fetal movement. At present, ultra-fast MRI, such as the HASTE sequence [5] can overcome this disadvantage. Moreover, the potential capability of MRI to image the pathological properties of the cartilaginous matrix may allow the differential diagnosis that is impossible solely on morphological grounds. Investigation of the optimal imaging sequence for the fetal skeleton should be encouraged. The combination of fetal MRI and US will be more effective for diagnosing skeletal dysplasias in utero.

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